DEPARTMENT OF HEALTH SERVICES

NEWBORN SCREENING PROGRAM 850 MARINA BAY PARKWAY, ROOM F175 RICHMOND, CA 94804 (510) 412-1502

ANY HOSPITAL **LABORATORY** 9876 First Avenue ANYTOWN, CA 99999

Newborn's Physician: JOHN SMITH, MD

Testing Laboratory:

WESTERN CLINICAL LABORATORY 408 SUNRISE AVE, ROSEVILLE, CA 95678

Directors:

John Sherwin, Ph.D., Chief,

Genetic Disease Laboratory, (510) 231-1790

NEWBORN SCREENING RESULTS - INITIAL

BABY

TestBaby

Baby's Birth:

Gender: Female NBS FORM #: 2000011132 Medical Record: ABC1234

BIRTH/COLLECTION INFORMATION

Date Time 07/19/05 0724 0115 Specimen Collection: 07/21/05

Age at Collection: 42 hour(s) Birth Weight: 2615 grams Ethnicity: White, Hispanic ANY HOSPITAL Specimen Collection Site:

Feeding Type: Breast only

MOTHER

TestBaby, Mom 123 Main St. Anytown, CA 99999 (800) 555-1212

Test interpretations are based on the **Birth/Collection Information** provided above and subject to disclaimer below.

| TEST - Individual analytes listed on reverse side | INTERPRETATION | |
|--|--|--|
| Galactosemia | Negative | |
| Primary Congenital Hypothyroidism | Negative | |
| Congenital Adrenal Hyperplasia (due to 21-Hydroxylase Deficiency) | Negative | |
| MS/MS Acylcarnitine Panel | Negative | |
| MS/MS Amino Acid Panel (Including PKU) | Negative | |
| Hemoglobinopathies | INTERPRETATION | |
| Hb Pattern: | Usual hemoglobin pattern. These results assume no transfusion prior to testing and do not rule out the possibility of a thalassemia trait or rare hemoglobin variants. | |

Follow-up:

Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider. If you have questions regarding these results, please contact the Newborn Screening staff at

Stanford University Medical Center, (650) 812-0353.

TestBaby, Female Date of Birth: 07/19/05

| Acylcarnitine | Reference Range | Result | Flag |
|----------------------------|----------------------|-----------------------|------|
| FC | 12 - 220 □mol/L | 18.4 | |
| FC / (C16 + C18:1) Ratio | 0 - 60 | 4.22 | |
| C-2 | 5 - 85 | 18.4 | |
| C-3 | 0 - 9.25 | 1.3 | |
| C03 / C02 Ratio | 0 - 0.25 | 0.07 | |
| C-3DC | | 0.120 | |
| C-4 | 0 - 2 | 0.220 | |
| C-4DC | 0 - 2.6 | 0.440 | |
| C-5 | 0 - 0.8 | 0.100 | |
| C-5:1 | 0 - 0.4 □mol/L | 0.000 | |
| C-5OH | 0 - 1.2 □mol/L | 0.240 | |
| C-5DC | 0 - 0.35 | 0.180 _mol/L | |
| C-6 | 0 - 0.7 □mol/L | 0.100 [mol/L | |
| C-8 | 0 - 0.5 | 0.100 [mol/L | |
| C08 / C10 Ratio | | 0.59 | |
| C-8:1 | 0 - 0.9 | 0.080 □m ol/ L | |
| C-10 | 0 - 0.6 | 0.170 | |
| C-10:1 | 0 - 0.45 | 0.080 [mol/L | |
| C-12 | | 0.300 | |
| C-12:1 | | 0.19 □mol/L | |
| C-14 | 0 - 1.1 | 0.260 | |
| C14:1 | 0 - 0.8 | 0.240 | |
| C14:1 / C12:1 Ratio | | 1.26 | |
| C-14OH | | 0.020 | |
| C-16 | 0 - 10 <u></u> mol/L | 3.170 | |
| C-16:1 | | 0.270 | |
| C-16OH | 0 - 0.3 [mol/L | 0.060 | |
| C-18 | 0 - 3.5 [mol/L | 1.040 | |
| C-18:1 | 0 - 4 □mol/L | 1.180 | |
| C-18:2 | | 0.16 | |
| C-18OH | 0 - 0.4 [mol/L | 0.000 | |
| C-18:1OH | 0 - 0.21 □mol/L | 0.080 | |

| Amino Acid | Reference Range | Result | Flag |
|------------------------------|-----------------|-----------|------|
| Glycine | 0 - 1800 | 620 | |
| Alanine | 0 - 900 | 311 | |
| Valine | | 114.2 | |
| Leucine/Isoleucine | 0 - 200 | 106.1 | |
| Leucine/Alanine Ratio | 0 - 1.25 | 0.34 | |
| Phenylalanine | 0 - 140 | 71.7 | |
| Phenylalanine/Tyrosine Ratio | 0 - 2.3 | 0.89 | |
| Tyrosine | 0 - 700 | 81.0 | |
| Methionine | 0 - 100 | 32 ∏mol/L | |
| Citrulline | 0 - 60 | 20 ∏mol/L | |
| Citrulline/Arginine Ratio | | 1.47 | |
| Ornithine | 0 - 450 | 102 | |
| Ornithine/Citrulline Ratio | | 5.12 | |
| Arginine | 0 - 200 | 14 □mol/L | |
| Arginine/Ornithine Ratio | | 0.13 | |
| Proline | 0 - 400 | 239 | |
| 5-Oxoproline | -1 - 9999.9999 | 71 □mol/L | |

| Analyte | Reference Range | Result | Flag |
|----------------------------------|-------------------|---------------------|------|
| Galactose -1 -uridyl transferase | > 50 enzyme units | 247.92 enzyme units | |
| TSH | 0 - 25 mIU/mL | 1.64 mIU/mL | |
| 17 Hydroxyprogesterone (17 OHP) | < 120 nmol/L | 0033.60 nmol/L | |